



TREX1 gene

three prime repair exonuclease 1

Normal Function

The *TREX1* gene provides instructions for making the 3-prime repair exonuclease 1 enzyme. This enzyme is a DNA exonuclease, which means it trims molecules of DNA by removing DNA building blocks (nucleotides) from the ends of the molecules. In this way, it breaks down unneeded DNA molecules or fragments that may be generated during copying (replication) of cells' genetic material in preparation for cell division, DNA repair, cell death, and other processes.

Health Conditions Related to Genetic Changes

Aicardi-Goutieres syndrome

At least 16 mutations in the *TREX1* gene have been identified in people with Aicardi-Goutieres syndrome. Most of these mutations are believed to prevent the production of the 3-prime repair exonuclease 1 enzyme. Researchers suggest that the absence of this enzyme may result in an accumulation of unneeded DNA and RNA in cells. These DNA and RNA molecules may be mistaken by cells for those of viral invaders, triggering immune system reactions that result in severe brain dysfunction (encephalopathy), skin lesions, and other signs and symptoms of Aicardi-Goutieres syndrome.

systemic lupus erythematosus

other disorders

Mutations in the *TREX1* gene have also been identified in people with other disorders involving the immune system. These disorders include a chronic inflammatory disease called systemic lupus erythematosus (SLE), including a rare form of SLE called chilblain lupus that mainly affects the skin.

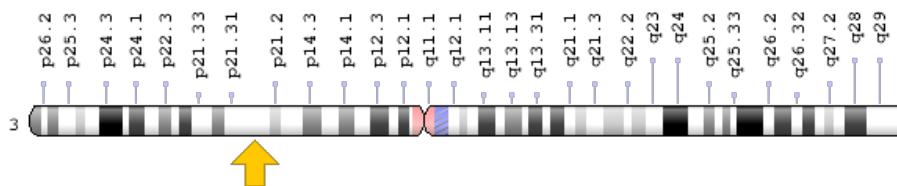
TREX1 gene mutations have also been found in people with a disorder called autosomal dominant retinal vasculopathy with cerebral leukodystrophy, which affects the brain and the blood vessels in the specialized light-sensitive tissue that lines the back of the eye (the retina).

As in Aicardi-Goutieres syndrome, absence or impaired function of the 3-prime repair exonuclease 1 enzyme may cause immune system dysfunction that damages the brain, skin, blood vessels, and other parts of the body in these conditions. It is not clear why mutations in the same gene cause several different disorders.

Chromosomal Location

Cytogenetic Location: 3p21.31, which is the short (p) arm of chromosome 3 at position 21.31

Molecular Location: base pairs 48,465,520 to 48,467,645 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 3'-5' exonuclease TREX1
- 3' repair exonuclease 1
- AGS1
- CRV
- deoxyribonuclease III, dnaQ/mutD-like
- DKFZp434J0310
- DNase III
- DRN3
- HERNS
- three prime repair exonuclease 1 isoform a
- three prime repair exonuclease 1 isoform b
- TREX1_HUMAN

Additional Information & Resources

GeneReviews

- Aicardi-Goutieres Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1475>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28TREX1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- 3-PRIME REPAIR EXONUCLEASE 1
<http://omim.org/entry/606609>
- CHILBLAIN LUPUS 1
<http://omim.org/entry/610448>
- VASCULOPATHY, RETINAL, WITH CEREBRAL LEUKODYSTROPHY
<http://omim.org/entry/192315>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_TREX1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TREX1%5Bgene%5D>
- HGNC Gene Family: Exonucleases
<http://www.genenames.org/cgi-bin/genefamilies/set/544>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=12269
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/11277>
- UniProt
<http://www.uniprot.org/uniprot/Q9NSU2>

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